รายงานผู้ป่วย

Marfan's syndrome with aneurysmal formation of the aortic root and ascending aorta concomitant with aortic valve regurgitation of patients undergoing Bentall operation: the first two cases in southern Thailand

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Abstract

Marfan's syndrome with aneurysmal formation of the aortic root and ascending aorta concomitant with aortic valve regurgitation of patients undergoing Bentall operation: the first two cases in southern Thailand

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The aorta is considered as pathologically dilated when the diameter of the aortic root and ascending aorta exceed the normal for a given age and body size. Such aneurysmal dilatation of the aortic root and ascending aorta frequently leads to significant aortic valve regurgitation.

Marfan's syndrome is a genetic disorder of connective tissue that is marked by abnormalities of the skeletal, ocular and cardiovascular system. The main cardiovascular features of Marfan's syndrome are progressive dilatation of the aortic root and ascending aorta, which may result in aortic rupture and aortic dissection or in aortic valvular regurgitation. The Bentall operation (one-piece composite valve-graft conduit for aortic valve, aortic root and ascending aorta replacement concomitant with coronary button reimplantation) is considered as a standard procedure to correct and prevent potential complications of this disease.

This is a report of the first two cases that had received Bentall operation in southern Thailand. The results of the surgical treatment was satisfactory.

Key words: *ascending aorta, aortic root, aneurysmal formation, aortic valve regurgitation, Marfan's syndrome, aortic rupture, aortic dissection, Bentall operation, valve-graft conduit, coronary button reimplantation.*

บทคัดย่อ:

หลอดเลือดแดงใหญ่เอออร์ตาที่มีขนาดของส่วน aortic root และส่วน ascending aorta ขยายใหญ่เกินกว่าระดับที่เหมาะสมกับอายุและสัดส่วนของร่างกาย ถือว่ามีพยาธิสภาพ และพยาธิสภาพแบบโป่งพอง (aneurysm) ของ aortic root และ ascending aorta มักเป็นสาเหตุทำให้เกิดภาวะลิ้นหัวใจเอออร์ติกรั่ว (aortic valve regurgitation) ร่วมด้วย

Marfan's syndrome เป็นความผิดปกติของระบบเนื้อเยื่อที่สามารถถ่ายทอดทางพันธุกรรม โดยมักมีความผิดปกติของระบบกล้ามเนื้อ, ระบบการมองเห็น รวมถึงระบบหัวใจและหลอดเลือด ลักษณะความผิดปกติของระบบหัวใจและหลอดเลือดที่สำคัญ คือ การขยายของ aortic root และ ascending aorta ในแบบโป่งพอง (aneurysm) ซึ่งอาจมีผลทำให้เกิดหลอดเลือดแดงใหญ่แตก (aortic rupture) ผนังหลอดเลือดด้านในฉีกและมีเลือดเซาะเข้าไปในผนังหลอดเลือด (aortic dissection) หรือลิ้นหัวใจเอออร์ติกรั่ว (aortic valve regurgitation) การผ่าตัดแบบ Bentall ซึ่งประกอบด้วยการเปลี่ยนลิ้นหัวใจเอออร์ติก การซ่อมและเปลี่ยน aortic root ร่วมกับ ascending aorta ด้วยหลอดเลือดเทียมและลิ้นหัวใจเทียมที่เย็บประกอบกันเป็นชิ้นเดียว (composite graft) ร่วมกับการตัดต่อหลอดเลือดหัวใจโคโรนารี (coronary artery) เข้ากับหลอดเลือดเทียม (coronary artery reimplantation) เป็นที่ยอมรับว่า เป็นวิธีการผ่าตัดที่แก้ไขความผิดปกติดังกล่าว และสามารถป้องกันผลแทรกซ้อนที่อาจเกิดขึ้นได้จากกลไกพยาธิสภาพของภาวะ Marfan ที่มีผลต่อหลอดเลือดแดงใหญ่เอออร์ตาได้

รายงานผู้ป่วยฉบับนี้เป็นรายงานผู้ป่วย 2 รายแรกที่ได้รับการผ่าตัดแบบ Bentall ในภาคใต้ของประเทศไทย และมีผลการผ่าตัดเป็นที่น่าพอใจ

คำสำคัญ: *หลอดเลือดแดงใหญ่เอออร์ตา, aortic root, พยาธิสภาพแบบโป่งพอง, ลิ้นหัวใจเอออร์ติกรั่ว, Marfan's syndrome, aortic rupture, aortic dissection, ลิ้นหัวใจเทียม, หลอดเลือดหัวใจโคโรนารี*

Introduction

In 1896, Antoine Marfan described a 5-year-old girl with extremely long limbs and characterized as having arachnid or spider like fingers.[1] This was the beginning of the elucidation of the Marfan syndrome as we know it today.

The Marfan syndrome is the most common genetically determined disorder affecting adults and occurs at a frequency of 1 per 10,000 births.[2] The genetic abnormality is the presence of mutations in the fibrillin gene located in the chromosome 15q.[3,4] Fibrillin is the core protein of microfibrils.

Fibrillin and elastin represent the major components of the elastic fiber system. This abnormality leads to the synthesis of a mutant fibrillin, inability to bind calcium and weakening of the elastic connective tissue.[4-6] Marfan's syndrome is an autosomal dominant disorder of connective tissue that is marked by abnormalities of the skeletal, ocular and cardiovascular systems.[1, 7, 8] The main cardiovascular features are progressive dilatation of the proximal aorta and annulus, which may result in aortic root dilatation, aortic aneurysm and aortic dissection or in aortic valvular incompetence and regurgitation.[8, 9] Histopathologically, the aorta demonstrates cystic medial degeneration, disruption of elastic fiber and fibrosis of the media.[10] Seventy-five to eighty-five percent of patients with Marfan's syndrome have aortic root dilatation with or without aortic regurgitation, and one third have significant mitral regurgitation.[11]

The average age of death is approximately 32 years.[12] The majority of deaths, 60 to 80 percent, result from cardiovascular complications related to aortic root dilatation.[12] The prognosis of Marfan's patients was formerly disappointing, and premature death was common.[8, 9, 13] This dismal outlook for patients with aortic root aneurysm changed dramatically with the new composite graft valve conduit operation introduced by Bentall and De Bono in 1968.[14]

The composite replacement of the aortic valve and ascending aorta is the standard operation for aortic root aneurysm in patients with Marfan's syndrome.[15] We report the first two cases of Marfan's syndrome patients with aortic root and ascending aorta aneurysm concomitant with aortic valve insufficiency who underwent the Bentall operation in Southern Thailand.

Case reports

Case 1

A 43-year-old Thai woman was referred to Songklanagarind Hospital on May 4, 2003 from Ranong Province Hospital with the chief complaint of progressive dyspnea and chest pain that migrated to her back, which she had suffered for 3 months. She had also developed slight palpitations two weeks previously. She experienced no orthopnea, no paroxysmal nocturnal dyspnea, no hoarseness, no diplopia, no blurred vision or other symptoms indicating severe congestive heart failure. The previous investigations from several physicians at the provincial hospital showed cardiomegaly and dilatation of the ascending aorta. The initial reason for referal was an ascending aortic aneurysm.

Physical examination revealed slightly long, thin limbs and fingers. The oral cavity examination revealed a high arch, doming palate. A mild to moderate degree of diastolic blowing murmur at the aortic valve area in the right second intercostal space anteriorly was noted. The blood pressure was 120 mmHg systolic pressure and 50 mmHg diastolic pressure. Her rhythm was 85 beats per minute and in sinus rhythm. At presentation, the patient was in New York Heart Association (NYHA) functional class II. Other system examinations were unremarkable. A chest radiographic study showed mild cardiomegaly and lobulated dilatation of the thoracic aorta (Fig 1). A computed tomography scan of the chest and abdomen showed diffuse dilatation of the ascending aorta (68x61 mm) and descending aorta (48x62 mm) down to the diaphragmatic hiatus level, no evidence of contrast leakage or extravasation or aortic ulcer, and a normal sized abdominal aorta (Fig 2). An electrocardiographic study showed normal sinus rhythm and normal ST-T segments. All laboratory investigations showed normal limits. An echocardiography examination showed aortic root dilatation, a moderate degree of aortic valve regurgitation, no regional wall motion abnormality and good left ventricular ejection fraction.

Indications for surgery in this patient were chronic aneurysm larger than 55 mm in diameter, aortic root dilatation and moderate aortic valve regurgitation in situation of Marfan's syndrome

Operative findings: chronic aneurysmal formation of the aortic root and ascending aorta extending to the proximal portion of aortic arch (Fig 3). Marked dilatation of the aortic valve annulus and poor coaptation of the aortic valve cusps. Both coronary ostia were normal.

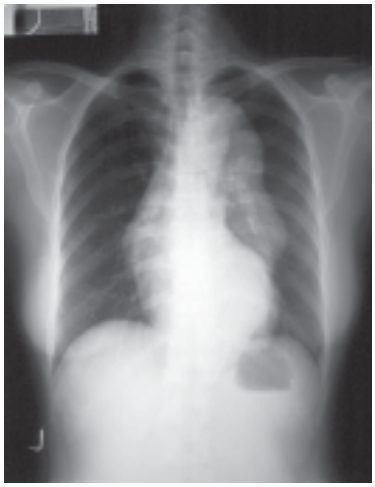

Fig 1 Chest radiograph of the patient from case report 1 with a large lobulated dilatation of the thoracic aorta

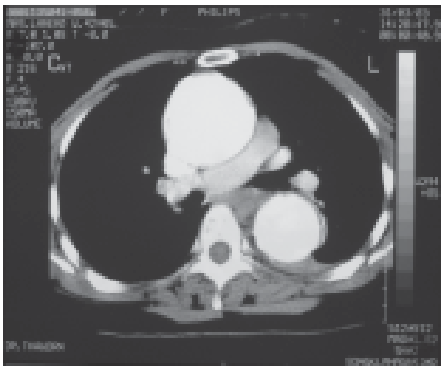

Fig 2 Computed tomographic image, enhanced by contrast medium, of a 6.8 cm. aneurysm of the ascending aorta

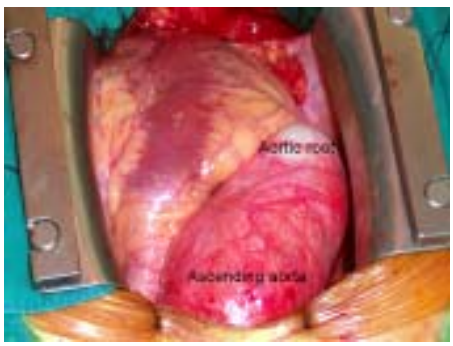

Fig 3 Intraoperative findings showing an aneurysmal formation of the aortic root, ascending aorta and proximal aortic arch

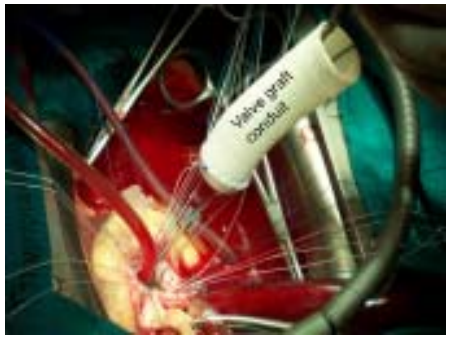

Fig 4 Intraoperative technique for aortic root replacement: placement of annular sutures to the valve conduit

Operative technique

Access was obtained through a median sternotomy. The femoral artery was cannulated and venous return was accomplished with a two-stage right atrial-inferior vena caval cannulation for performing a cardiopulmonary bypass. After cold cardiac arrest was obtained, the aneurysm was resected, including the aortic valve cusps, aortic root, sinus of valsalva and ascending aorta. The two coronary ostia were excised with a button technique. A composite, zero-porosity Dacron graft with a presewn prosthetic valve (size 21 mm in diameter) was secured with a continuous suture of 3-0 monofilament polypropylene. During the procedure, the patient was perfused with blood cooled to 12°C until the body temperature (nasopharyngeal) reached 16 to 18°C. A Bentall procedure (aortic valve and aortic root replacement with composite valve-graft conduit and reimplantation of the coronary arteries using the button technique) was performed (Fig 4). Next time, total body circulatory arrest was initiated under conditions of 16°C body temperature for distal anastomosis of the graft conduit to the unclamped aortic arch, because clamping the aorta in this area would occlude cerebral blood flow. After the repair was completed, the body was rewarmed to normothermia. The patient was discontinued from the cardiopulmonary bypass in the usual manner for complete operation. Total operative time, aortic clamp time, cardiopulmonary bypass time and circulatory arrest time were 420, 124, 291 and 18 minutes respectively. There were no immediate postoperative complications.

The patient was extubated within 12 hours postoperatively. The intensive care unit stay was 35 hours. The first postoperative and most recent echocardiographic studies showed good aortic valve prosthetic function and good left ventricular function. The total hospital stay was 8 days. At present, one year postoperatively, the patient is healthy and is NYHA functional class I. The result of surgical treatment was satisfactory.

Case 2

A 24-year-old Thai man was referred to Songklanagarind Hospital on August 29, 2003 from Nakornsritammarat Provincial Hospital with the chief complaint of progressive dyspnea for sixteen months. He was in NYHA functional class IV. He was unable to do any activities or jobs except a little self care in his daily life. He had received diuretic drug and digoxin for six months, but experienced little improvement.

The physical examination revealed markedly long, thin limbs and fingers. He had lens subluxation and a very high arch doming palate. Blood pressure was 140 mmHg systolic pressure and 40 mmHg diastolic pressure. His rhythm was 92 beats per minute and in normal sinus rhythm. He had typical signs of severe aortic regurgitation, revealed by auscultatory sounds and a precordium examination. The chest radiographic study showed marked cardiomegaly and left ventricular chamber enlargement, and dilatation of the ascending aorta (Fig 5). A computed tomography scan of the chest showed fusiform dilatation of the ascending aorta and aortic root, with maximum diameter at the aortic root level of 90x 92 mm (Fig 6). A transesophageal echocardiography examination showed aortic root dilatation and ascending aortic aneurysm, normal size aortic arch and descending aorta, severe aortic regurgitation, and left ventricular ejection fraction 40 percent. After receiving consultation from the cardiology unit, We decided to operate for correction of these problems as soon as possible, as we regarded this as an urgent situation.

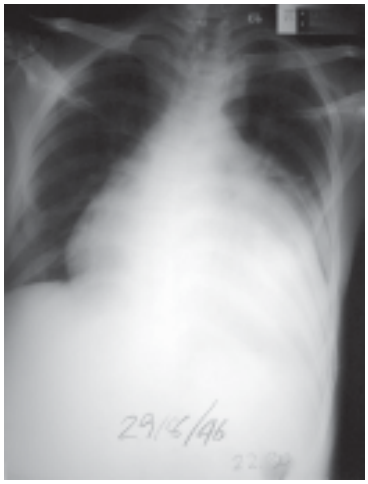

Fig 5 Chest radiograph of the patient from case report 2 showing dilatation of the ascending aorta and left ventricle

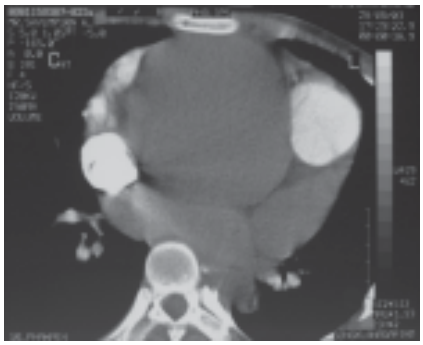

Fig 6 Computed tomographic image showing a large aneurysmal formation of the ascending aorta and aortic root

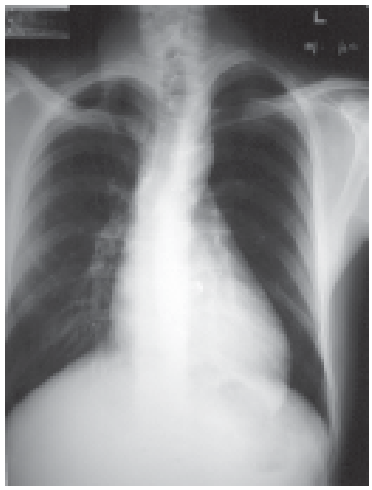

Fig 7 Postoperative chest radiograph of the patient from case report 2 showing normal cardiac size

Operative technique

For this patient, all operative techniques were the same as for the previously discussed patient, except that operation for circulatory arrest was not performed because the distal portion of the ascending aorta was free of aneurysms. The operative time, aortic clamp time and cardiopulmonary bypass time were 605, 155 and 284 minutes respectively. Immediately postoperatively, the patient had cardiac tamponade and cardiac arrest. An emergency median sternotomy was performed in the intensive care unit with cardiopulmonary resuscitation until the patient returned to hemodynamic stability. Reclosure of the sternotomy was done in the intensive care unit under sterile techniques. The patient was extubated within 12 hours postoperatively. The intensive care unit stay was 48 hours. The patient developed postpericardiotomy syndrome in the form of massive pericardial effusion in the 12th postoperative day and received subxiphoid pericardial drainage. A postoperative chest radiographic study showed a decrease in cardiac size (Fig 7). The first postoperative and most recent echocardiographic studies showed good aortic valve prosthetic function, improved left ventricular function to 55 percent and no pericardial effusion. The total hospital stay was 19 days. At present, eight months postoperatively, the patient is healthy, has better exercise tolerance and is NYHA functional class I. The result of surgical treatment was satisfactory.

Discussion

Without surgery, most patients with Marfan's syndrome have a dismal prognosis and die in the third decade of life from cardiovascular complications, such as aortic aneurysm rupture, aortic dissection or aortic regurgitation.[8, 16] Aortic root replacement with composite valve-graft dramatically improves the survival of Marfan's syndrome patients.[15] In a recent report by Gott and associates[17] on the results of aortic root replacement in 271 patients with Marfan's syndrome, there was no operative mortality among 235 patients operated on electively and operative mortality was only 5.6 percent among 36 patients receiving urgent operations. Successful employment of the Bentall operation (composite valve-graft replacement and coronary reimplantation) is considered to have been the single most important factor responsible for improvement in the prognosis and near normal life expectancy of these patients.[7, 9, 15, 18]

In our two cases reported here, a long operative time resulted from the time using circulatory arrest process and because it was our first experience with this procedure. For the second case, we hoped to improve the operative strategies, but the operative time, aortic clamp time and cardiopulmonary bypass times were longer than in the first case because a lot of time was used to stop bleeding at the neoaortic root, which had resulted from a less than optimal decision regarding an appropriate valve-graft conduit size.

A recent survey of 10 major Marfan surgical centers worldwide reported an operative mortality rate of 1.5 percent for Marfan's patients undergoing elective repair of aortic root aneurysm.[15] Thromboembolism and endocarditis are the most common late complications occuring after a Bentall procedure. Aortic valve sparing procedures[19, 20] have emerged as an alternative to composite valve-graft aortic root replacement, and avoiding the need for an anticoagulant and late valve-related complications may benefit children, women of childbearing age and patients in whom administration of anticoagulants is contraindicated. The current recommendation is to operate when the size of the aortic root reaches 55 to 60 mm[15] for prophylaxis of catastrophic aortic events in Marfan's patients.

Conclusions

In Marfan's syndrome patients, elective aortic root replacement, usually for a chronic aortic root aneurysm, can be performed with a low operative risk, low operative mortality and good late survival. Early prophylactic surgery is recommended for prevention of catastrophic aortic events. Long-term clinical and chest radiographic follow-up are needed in all operated patients. Finally, the author and surgical teams and our institute have a strong commitment to develop better operative strategies for the Bentall procedure in Marfan and non-Marfan patients for lowering morbidity, mortality and improving the patients' quality of life.

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